Behind the Screens

Missouri Department of Health and Senior Services
Newborn Screening Program

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Featured Disorder

Galactosemia is a rare, genetic, metabolic disorder in which those affected are unable to properly digest the sugar galactose. Galactose is one of the main components of lactose, the primary sugar found in milk and dairy products, including breastmilk. If a baby with galactosemia consumes galactose, undigested sugars build up in the blood, rather than being used for energy. If galactosemia is left untreated, it can cause seizures, serious blood infections, liver damage, or even death. There are multiple forms of galactosemia, but newborn screening primarily targets classic galactosemia, as it can affect babies within days of birth.

Classic galactosemia affects 1 in every 30,000 to 60,000 newborns. It occurs in people of all ethnic groups, but is most common in those of Irish descent. Some of the early signs of galactosemia include: poor feeding and sucking, vomiting, diarrhea, irritability and low blood sugar. If galactosemia is identified early in life, through newborn screening, intervention and proper treatment can begin immediately. Treatment for galactosemia includes working with specialists and dieticians to ensure the child maintains a lactose-free diet. If treatment is started after 10 days of life, delays or learning problems are more likely. The level of delay varies from child to child. Treatment is still important, even if started late, because it can help prevent further delays and symptoms. If proper treatment is begun immediately, children with galactosemia often can lead healthy lives.

Each year Missouri's Newborn Screening Program identifies children with multiple forms of galactosemia. With your help of collecting and transporting newborn screening specimens in a timely manner, the state can help ensure that affected children receive early intervention, giving them the best opportunity to lead a normal life.

What's New?

The DHSS is now collecting baby level critical congenital heart disease (CCHD) screening results for all babies born in Missouri. This is a recent change from the voluntary aggregate reporting that has been collected since the implementation of statewide CCHD screening in January 2014. RSMo 191.334, otherwise known as Chloe's Law, requires that all babies born in Missouri be screened for CCHD. In addition to screening all newborns for CCHD, Chloe's Law also requires that screening results shall be reported to the DHSS in a manner prescribed by the DHSS for surveillance purposes.

This past fall, the DHSS filed the final rules for CCHD screening, 19 CSR 40-12.010, with the Secretary of State and the rules became effective November 30, 2017. The rules establish screening guidelines and provide direction for reporting of screening results and will replace the voluntary aggregate reporting system. For more information on CCHD reporting please visit www.health.mo.gov/cchd.

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Did You KNOW?

Did you know that the Missouri State Public Health Laboratory (MSPHL) tests newborn blood spot screening samples six days per week including Saturdays and most holidays? In 2014, the MSPHL began implementing changes to improve newborn blood spot screening transit time. Transit time is the time from sample collection to the start of testing by the MSPHL. Minimizing transit time is crucial to ensure screening results are reported promptly to allow for identification and intervention for time-critical disorders. To achieve transit time improvement, the courier service that transports the samples to the MSPHL was expanded to include Sunday and holiday pickup. In addition to the courier enhancements, the MSPHL also expanded their workweek from five to six days and began testing on most holidays. The MSPHL now tests blood spot samples Monday through Saturday, every week of the year. The only holidays that testing and courier service does not occur are Thanksgiving Day, Christmas Day and New Year's Day. These changes, in addition to the efforts of submitters, have resulted in an improvement in timeliness from 61 percent of samples being received within three days of collection in 2013 to 90 percent in 2016.

Tech Tips

- Bright or infrared light, including bilirubin lamps and surgical lights, can affect the accuracy of a pulse ox reading. Ensure that the infant is not placed in bright or infrared light while critical congenital heart disease (CCHD) screening is being performed. You may cover the pulse ox sensor with a blanket to ensure that extraneous light does not affect the accuracy of your reading.

- It is important to identify the health care provider who will care for the newborn after discharge for all newborn screenings. To document this information in MoEVR, use the free-text box and provide the health care provider's first and last name. A clinic name or office phone number will also suffice.

- Never throw away a newborn screening (NBS) collection form. If you make a mistake or if you suspect the specimen will be a poor quality sample, allow the specimen to dry completely, write VOID across the front of the form and return it to the State NBS Laboratory along with the correct form. The voided form will be replaced free of charge.

PATIENT SPOTLIGHT

Braxton

Braxton is a Missouri child living with congenital hypothyroidism (CH). Without early detection and intervention, children with CH can be affected by slow growth, poor weight gain or learning delays. Braxton's screen was collected, transported, and tested in an appropriate time frame which has given him the best start for a happy, healthy life. His mother, Megan, said this about their experience:

"My husband and I welcomed our second sweet baby boy, Braxton, into this world on June 29, 2016. Being our second delivery we thought things would be just like the first. We were released home on a Friday afternoon. We were settling in nicely as a family of 4, when the phone rang not even 12 hours after being home. It was the pediatrician. The pediatrician told us we needed to get to the hospital as soon as possible. Braxton's thyroid levels came back elevated on his newborn screening and he needed additional testing. Depending on these tests we may be readmitted to the hospital. The 45 minute drive to the hospital was what felt like the longest drive of my life. A series of blood draws were done. When the pediatrician called us after the results were back, she had talked with the endocrinologist in Columbia, MO, and we were told we could go back home and follow up with the endocrinologist the following week. We followed up as instructed. More labs were taken and Braxton was then diagnosed with congenital hypothyroidism. His thyroid was producing thyroid hormone but not an adequate amount to maintain a normal level. He was started on medication when he was less than 2 weeks old. We continue to see the endocrinologist for lab work and medication management. Without the newborn screening we may have never known of Braxton's condition until it was too late. We are forever grateful for early detection and treatment interventions to help ensure normal growth and development!

> "Without the newborn screening we may have never known of Braxton's condition until it was too late." *Megan*

THANK YOU for your contribution to ensuring the best possible start for Missouri newborns.

Missouri DHSS Department of Health and Senior Services

MISSOURI DEPARTMENT OF
HEALTH AND SENIOR SERVICES

Bureau of Genetics and Healthy Childhood
Newborn Blood Spot, Hearing, and CCHD Programs
www.health.mo.gov/newbornscreening
573.751.6266 or 800.877.6246

Missouri State Newborn Screening Laboratory
573.751.2662